Early presentation of endomyocardial fibrosis

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Dear Dr Baker,

Having read the interesting report by Cilliers et al about a patient with early presentation of endomyocardial fibrosis at the age of 22 months,¹ we would like to make some comments in comparison with one case we reported some years ago.² Certainly, Cilliers’ case is one of the youngest already reported in the literature. They state that the youngest patient reported was an 18-month-old baby who had the diagnosis based on echocardiographic findings.³ However, our patient was younger (a 4-month-old Caucasian baby) and we also provided histological confirmation of the disease. In fact, we believe that our patient presented the cardiac lesions since birth, although they were not clinically recognised at that time. The north-eastern region of Brazil, like some African countries, is considered an endemic area for endomyocardial fibrosis, but neither the baby nor his parents were originally from that area. Additional features in our case were the existence of Rh incompatibility between the mother and foetus and treated hypothyroidism in the mother.

We agree with the authors regarding a possible genetic link in the aetiology of the disease, but we additionally believe that congenital development of the cardiac lesions may exist. The study of these ‘extremely early’ presentation forms may help in the understanding of the pathogenetic mechanisms of endomyocardial fibrosis, which are still not completely understood.

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References